The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000314.8(PTEN):c.334C>G (p.Leu112Val)

Version: 1.0

CA377482212

1406308 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 6078cf83-90b3-48a9-b033-c55d588185c3
Approved on: 2024/4/5
Published on: 2024/4/10

HGVS expressions

NM_000314.8:c.334C>G
NM_000314.8(PTEN):c.334C>G (p.Leu112Val)
NC_000010.11:g.87933093C>G
CM000672.2:g.87933093C>G
NC_000010.10:g.89692850C>G
CM000672.1:g.89692850C>G
NC_000010.9:g.89682830C>G
NG_007466.2:g.74655C>G
ENST00000700029.2:c.334C>G
ENST00000710265.1:c.334C>G
ENST00000472832.3:c.334C>G
ENST00000688158.2:n.1069C>G
ENST00000688922.2:c.*164C>G
ENST00000700021.1:c.289C>G
ENST00000700022.1:c.334C>G
ENST00000700029.1:c.168C>G
ENST00000706954.1:c.334C>G
ENST00000706955.1:c.*369C>G
ENST00000686459.1:c.334C>G
ENST00000688158.1:c.*445C>G
ENST00000688308.1:c.334C>G
ENST00000688922.1:c.255C>G
ENST00000693560.1:c.853C>G
ENST00000371953.8:c.334C>G
ENST00000371953.7:c.334C>G
ENST00000498703.1:n.160C>G
ENST00000610634.1:c.232C>G
NM_000314.5:c.334C>G
NM_000314.6:c.334C>G
NM_001304717.2:c.853C>G
NM_001304718.1:c.-417C>G
NM_000314.7:c.334C>G
NM_001304717.5:c.853C>G
NM_001304718.2:c.-417C>G

Likely Pathogenic

Met criteria codes 4

PM2_Supporting PP3 PS4_Moderate PS3

Not Met criteria codes 2

PP2 BS3

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specification: *ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.1.0*

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

NM_000314.8(PTEN):c.334C>G (p.Leu112Val) meets criteria to be classified as likely pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.1.0). Please see a summary of the rules and criteria codes in the "PTEN ACMG Specifications Summary" document (assertion method column). PS3: Phosphatase activity <50% of wild type OR RNA, mini-gene, or other assay shows impact on splicing. (internal laboratory contributor). PS4_M: Proband(s) with phenotype specificity score of 2-3.5. (PMID: 28677221). PM2_P: Absent in gnomAD. PP3: SpliceAI predicts a splicing impact.

Met criteria codes

PM2_Supporting	✔	Absent in gnomAD.
PP3	✔	SpliceAI predicts a splicing impact.
PS4_Moderate	✔	Proband(s) with phenotype specificity score of 2-3.5. (PMID: 28677221).
PS3	✔	Phosphatase activity <50% of wild type OR RNA, mini-gene, or other assay shows impact on splicing. (internal laboratory contributor: SCV002606143.1): RNA analysis was completed. An exon skipping partial (ESP) event was seen to be associated with this variant: r.334_492del (p.Leu112_Lys164del). And the ESP event was complete. By manual view in IGV, 100% of reads that support normal splicing contain the wild type allele.

Not Met criteria codes

PP2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3	✖	Miguell cumulative score, -0.223486317. (WT-like)

Curation History

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